



DEPARTMENT OF COMMERCE

National Institute of Standards and Technology

Genome in a Bottle Consortium – Progress and Planning Workshop

AGENCY: National Institute of Standards & Technology (NIST), Commerce.

ACTION: Notice of public workshop.

SUMMARY: NIST announces the Genome in a Bottle Consortium meeting to be held on Thursday and Friday, January 29 and 30, 2015. The Genome in a Bottle Consortium is developing the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls. A principal motivation for this consortium is to enable performance assessment of sequencing and science-based regulatory oversight of clinical sequencing. The purpose of this meeting is to update participants about progress of the consortium work, continue to get broad input from individual stakeholders to update or refine the consortium work plan, continue to broadly solicit consortium membership from interested stakeholders, and invite members to participate in work plan implementation. Topics of discussion at this meeting will include release of the pilot candidate NIST Reference Material, short and long read data from the next sets of NIST Reference Materials, structural variants, and potential future Reference Materials.

DATES: The Genome in a Bottle Consortium meeting will be held on Thursday, January 29, 2015 from 9:00 AM to 5:30 PM Pacific Time and Friday, January 30, 2015 from 9:00 AM to 12:45 PM Pacific Time. Attendees must register by 5:00 PM Pacific Time on Thursday, January 22, 2015.

ADDRESSES: The meeting will be held in the Fisher Conference Center inside the Arrillaga Alumni Center at Stanford University, 326 Galvez Street, Stanford, CA 94305-6105. Please note admittance instructions under the SUPPLEMENTARY INFORMATION section of this notice.

FOR FURTHER INFORMATION CONTACT: For further information contact Justin Zook by email at jzook@nist.gov or by phone at (301) 975-4133 or Marc Salit by email at salit@nist.gov or by phone at (650) 350-2338. To register, go to: <http://app.certain.com/profile/form/index.cfm?PKformID=0x19409085b04>

SUPPLEMENTARY INFORMATION: Clinical application of ultra high throughput sequencing (UHTS) for hereditary genetic diseases and oncology is rapidly growing. At present, there are no widely accepted genomic standards or quantitative performance metrics for confidence in variant calling. These standards and quantitative performance metrics are needed to achieve the confidence in measurement results expected for sound, reproducible research and regulated applications in the clinic. On April 13, 2012, NIST convened the

workshop “Genome in a Bottle” to initiate a consortium to develop the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls (www.genomeinabottle.org). On August 16-17, 2012, NIST hosted the first large public meeting of the Genome in a Bottle Consortium, with about 100 participants from government, academic, and industry. This meeting was announced in the Federal Register (77 FR 43237) on July 24, 2012. A principal motivation for this consortium is to enable science-based regulatory oversight of clinical sequencing.

At the August 2012 meeting, the consortium established work plans for four technical working groups with the following responsibilities:

- (1) Reference Material (RM) Selection and Design: select appropriate sources for whole genome RMs and identify or design synthetic DNA constructs that could be spiked-in to samples for measurement assurance.
- (2) Measurements for Reference Material Characterization: design and carry out experiments to characterize the RMs using multiple sequencing methods, other methods, and validation of selected variants using orthogonal technologies.
- (3) Bioinformatics, Data Integration, and Data Representation: develop methods to analyze and integrate the data for each RM, as well as select appropriate formats to represent the data.
- (4) Performance Metrics and Figures of Merit: develop useful performance metrics and figures of merit that can be obtained through measurement of the RMs.

The products of these technical working groups will be a set of well-characterized whole genome and synthetic DNA RMs along with the methods (documentary standards) and reference data necessary for use of the RMs. These products will be designed to help enable translation of whole genome sequencing to regulated clinical applications. The consortium meets in workshops two times per year, in January at Stanford University in Palo Alto, CA, and in August at the National Institute of Standards and Technology in Gaithersburg, MD. At these workshops, including the last meetings at Stanford in January 2014 and at NIST in August 2014, participants in the consortium have discussed progress developing well-characterized genomes for NIST Reference Materials and planned future experiments and analysis of these genomes (see <https://federalregister.gov/a/2012-18064>, <https://federalregister.gov/a/2013-18934>, and <https://federalregister.gov/a/2014-18841> for past workshops at NIST). The August 2014 meeting, which included meetings of each of the four working groups, was announced in the Federal Register (79 FR 46774) on August 11, 2014, and the meeting is summarized at <https://sites.stanford.edu/abms/content/workshop-summary-and-slides-august-2014>.

There is no cost for participating in the consortium. No proprietary information will be shared as part of the consortium, and all research results will be in the public domain.

All attendees are required to pre-register. Anyone wishing to attend this meeting must pre-register at <http://app.certain.com/profile/form/index.cfm?PKformID=0x19409085b04> by 5:00 PM Pacific Time on Thursday, January 22, 2015, in order to attend.

Dated: January 14, 2015.

Richard Cavanagh
Acting Associate Director of Laboratory Programs

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